



Government of **Western Australia**
Department of **Health**



Royal Perth Hospital

Familial Hypercholesterolaemia

A Guide for Patients and their Families



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What is cholesterol?

Cholesterol is a fatty substance that the body needs to make hormones, vitamin D and to aid digestion.

Cholesterol is also needed to build and maintain the outer wall of cells. The body makes cholesterol and many of us consume some in our food.

How is cholesterol moved around the body?

The cholesterol from our diet is absorbed into the bloodstream and small particles called lipoproteins carry cholesterol around the body. Low-density lipoproteins (LDL) move cholesterol into the cells. The body's cells take up the LDL cholesterol via an LDL receptor. Liver cells take up most LDL cholesterol.

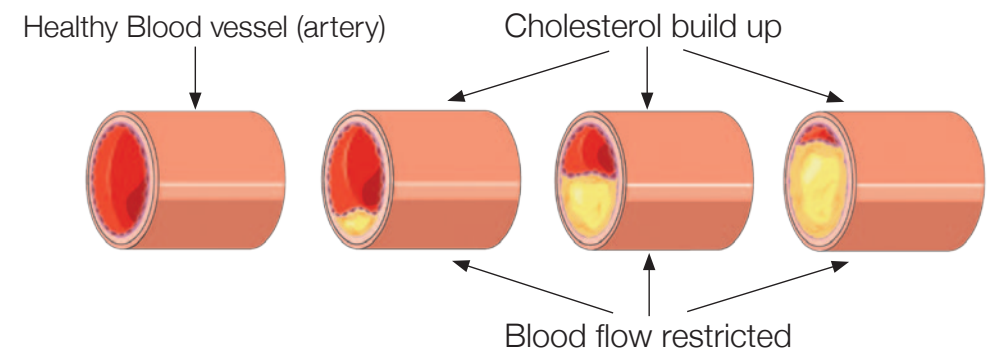
This controls the amount of cholesterol in the blood.

High-density lipoprotein (HDL) takes cholesterol out of cells and carries it to the liver. From here it leaves the body via the intestines. Both HDL and LDL cholesterol are needed for good health.

Can cholesterol be harmful?

If there is too much cholesterol in the bloodstream, it can deposit on the walls of the blood vessels. As shown in Figure 1, this causes the blood vessels to narrow and they may eventually become blocked.

Figure 1: High blood cholesterol leads to cholesterol build up in a blood vessel wall.



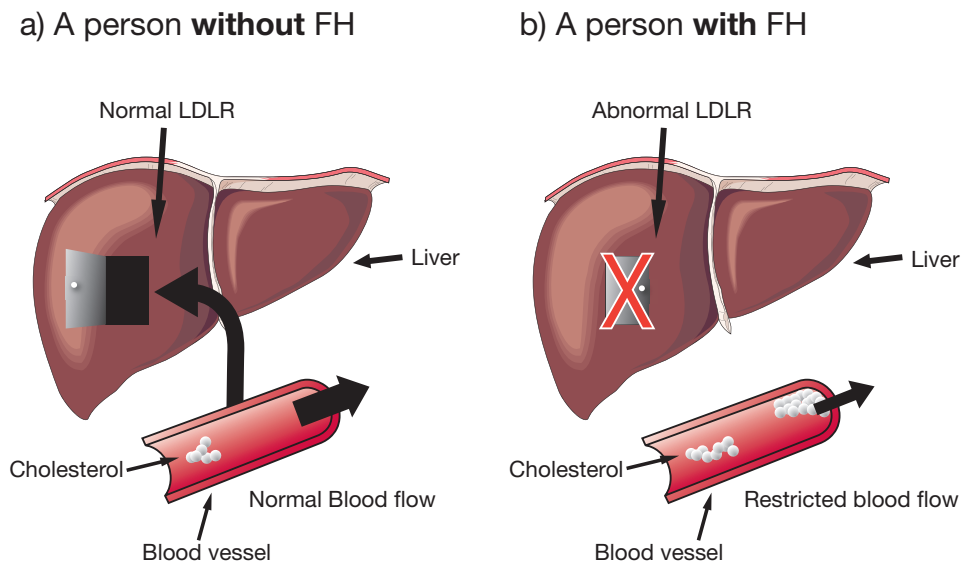
Courtesy of Servier Medical Art

This will affect blood flow to the heart and other vital organs and can cause heart attacks, strokes and other problems. A poor diet, diabetes, lack of exercise and obesity all increase the risk of high cholesterol and heart disease.

What is familial hypercholesterolaemia?

Some people have an increased risk of high cholesterol and heart disease due to an inherited condition known as familial hypercholesterolaemia (FH). This literally means “high cholesterol in a family”. In Australia, about 1 person in 500 people has FH. For people with FH the LDL receptors in the liver cells do not work properly and can not remove LDL cholesterol from the bloodstream efficiently.

An LDL receptor can be thought of as a door that allows the LDL cholesterol to be removed from the blood by the liver (Fig. 2a). In people with FH this door is only half open or, in severe cases, may be closed (Fig. 2b). This means the LDL cholesterol builds up in the bloodstream and causes the blood vessel to become blocked (Fig 2).



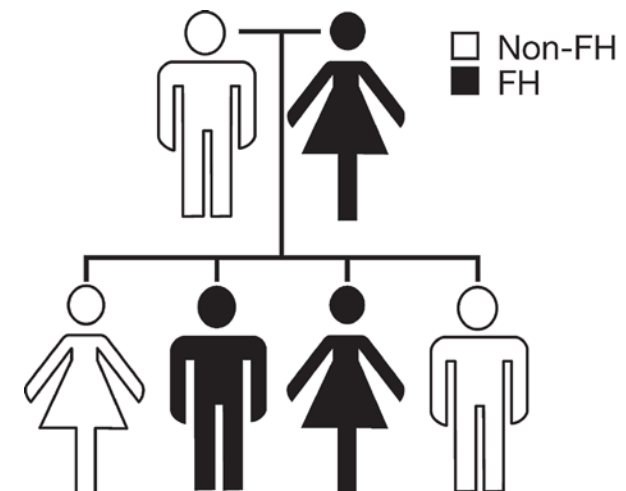
How do you inherit FH?

Everybody inherits one copy of the LDL receptor gene from their father and one from their mother. Genes are like an ‘instruction manual’ that tell your body how things should work.

In the case of FH, one or both of the genes inherited has an alteration ‘in the instruction manual’. This changes the way the LDL receptor works. If one gene is altered, the person has heterozygous FH, the most common form of the disease. If both genes are altered, the person has homozygous FH. Homozygous FH is very rare and more severe.

If a person has been diagnosed with heterozygous FH (and if their partner does not have FH), their children will have a 1 in 2 (or 50%) chance of inheriting the altered LDL receptor gene and having heterozygous FH (Fig. 3)

Figure 3: The inheritance of FH



What can be done about FH?

Many people born with FH may be able to avoid early onset heart disease by leading a healthy lifestyle along with taking medications prescribed by their doctor. A healthy lifestyle includes eating a healthy diet, not smoking, exercising regularly, staying at a healthy weight and enjoying alcohol in moderation.

Could I have FH?

FH runs in families, so it is useful to know the health of close family members (parents, brothers, sisters and children). If a close family member had heart disease at an early age (before 60 years) this may suggest increased risk of FH. Knowing if any of your relatives has high cholesterol may also be helpful.

If more than one close relative has early heart disease or high cholesterol, you may be at greater risk of having FH.

In certain groups of people, such as Afrikaaner, Christian Lebanese, Tunisian, French-Canadian or Ashkenazi Jew, the incidence of FH may be higher still. It is estimated that a large number of people with FH in Australia have not been diagnosed as having the condition.

What should I do?

If you have concerns about your own, or your family's cholesterol levels, you should see your general practitioner. A small sample of your blood can be taken and the cholesterol levels measured. Your doctor may also do a simple examination and ask for details of your family history. You may be referred to a specialist clinic that deals with cholesterol disorders for further tests, including genetic testing.

For more information contact:

Lipid Disorders Clinic

Cardiology
4th Floor South Block
Royal Perth Hospital
Ph: (08) 9224 8092

FH Family Support Group

www.fhfamilysupportgroup.websyte.com.au

Quit WA

www.quitwa.com or Quitline: 13 QUIT (13 7848)

National Heart Foundation

www.heartfoundation.org.au

Heart UK

www.heartuk.org.uk

Notes

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